Chronic Progressive External Ophthalmoplegia

CPEO

Chronic Progressive External Ophthalmoplegia, also known as CPEO, is a symptom that can occur in several mitochondrial conditions. Mitochondria are parts of a cell that help turn the energy we get from food into energy that the body can use. They are also important in the communication between body parts and creating other materials the body needs. Mitochondrial conditions can cause a variety of signs and symptoms in many parts of the body, particularly those that use a lot of energy like muscles and the brain.

Genetics

CPEO can be part of several genetic conditions. These can be due to changes in either the nuclear DNA, a set of DNA present in every cell of the body and inherited from both parents, or the mitochondrial DNA, a set of DNA contained in the mitochondria of a cell. The way the condition is passed down in families, called inheritance, will depend on the exact gene involved for the person with CPEO. Both males and females can have CPEO.

Frequency

The exact frequency of CPEO is unknown and likely underdiagnosed.

Signs and Symptoms

CPEO involves a loss of muscle function in the eye and eyelid. Features usually begin between the ages of 18 to 40 years.

These commonly include:

- Weakness or paralysis (inability to move) of the muscles that move the eye called ophthalmoplegia
- Drooping of the eyelids known as ptosis

CPEO can occur by itself, called an isolated feature, or present with other signs and symptoms, called a syndromic presentation. Individuals who are living with CPEO and present with other signs and symptomss are sometimes said to have **“CPEO plus.”**

Other signs and symptoms can include:

- Muscle pain or weakness called myopathy, especially during exercise
- Problems with muscle coordination (ataxia) or tremors (parkinsonism)
- Difficulty swallowing
- Hearing loss caused by nerve damage in the inner ear called sensorineural hearing loss
- A feeling of “pins and needles” or loss of feeling in limbs called neuropathy
- Depression or other mental health conditions

Treatment and Management

Before beginning any treatment or therapy, please consult with your physician.

As of 2022, there is currently no FDA-approved therapy for CPEO. However, there are treatment and management options related to the symptoms of CPEO.
This may include:

- Surgery for ptosis or strabismus if double vision occurs
- Special glasses that have a “ptosis crutch” to lift the upper eyelid
- Mitochondrial supplements like coenzyme Q10 in some cases
- Avoidance of mitochondrial toxins like certain drugs, tobacco, and alcohol

Individuals living with CPEO typically see an eye doctor called an ophthalmologist regularly. If someone has CPEO and additional symptoms (CPEO plus), they may follow up with additional doctors like neurology for their muscles and brain.

Clinical Trials

For specific details on clinical trials visit www.mitoaction.org/clinicaltrials or www.clinicaltrials.gov.

Resources

- Chronic progressive external ophthalmoplegia - About the Disease – The National Institute of Health
- Chronic progressive external ophthalmoplegia - NORD (National Organization for Rare Disorders) – The National Organization for Rare Disorders

Connecting with others who are impacted by a rare disease allows for important information to be shared about day-to-day life, prevents isolation, and gives hope. Please contact MitoAction for peer support opportunities at 888-MITO-411 or email mito411@mitoaction.org.

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